



## ATXN2 gene

ataxin 2

### Normal Function

The *ATXN2* gene provides instructions for making a protein called ataxin-2. This protein is found throughout the body, but its function is unknown. Ataxin-2 is found in the fluid inside cells (cytoplasm) and seems to interact with a cell structure called the endoplasmic reticulum. The endoplasmic reticulum is involved in protein production, processing, and transport. Researchers believe that ataxin-2 may be involved in processing RNA, a chemical cousin of DNA. Ataxin-2 is also thought to play a role in the translation of genetic information to produce proteins.

One region of the *ATXN2* gene contains a DNA segment known as a CAG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (cytosine, adenine, and guanine) that appear multiple times in a row. Normally, the CAG segment is repeated approximately 22 times within the gene.

### Health Conditions Related to Genetic Changes

amyotrophic lateral sclerosis

spinocerebellar ataxia type 2

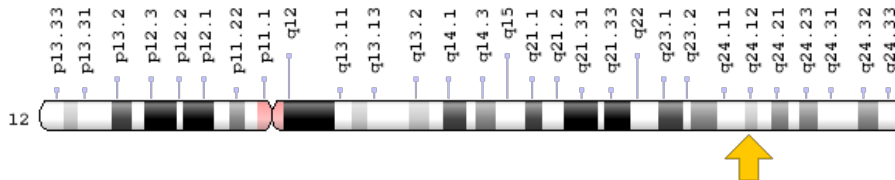
Spinocerebellar ataxia type 2 (SCA2) is a condition characterized by progressive problems with movement. SCA2 results from a mutation in the *ATXN2* gene known as a trinucleotide repeat expansion. This mutation increases the length of the repeated CAG segment in the *ATXN2* gene. People with 32 or more repeats CAG repeats in the *ATXN2* gene develop SCA2.

It is unclear how the abnormally long CAG segment affects the function of the ataxin-2 protein. The abnormal protein apparently leads to cell death, as people with SCA2 show a loss of brain cells. Certain brain cells called Purkinje cells seem to be particularly sensitive to the presence of abnormal ataxin-2. Purkinje cells are located in the part of the brain that coordinates movement (cerebellum) and are involved in chemical signaling between nerve cells (neurons). It is unknown how the abnormal ataxin-2 protein leads to the death of Purkinje and other brain cells. Over time, the loss of these cells causes the movement problems characteristic of SCA2.

## Chromosomal Location

Cytogenetic Location: 12q24.12, which is the long (q) arm of chromosome 12 at position 24.12

Molecular Location: base pairs 111,452,214 to 111,599,676 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ataxin-2
- ATX2
- ATX2\_HUMAN
- SCA2

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Trinucleotide expansion is one of the most important categories of mutation underlying neurodegenerative disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK27942/#A2851>
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/ataxia/domatax.html#sca2>

### GeneReviews

- Spinocerebellar Ataxia Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1275>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ATXN2%5BTIAB%5D%29+OR+%28ataxin+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- ATAXIN 2  
<http://omim.org/entry/601517>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ATXN2.html](http://atlasgeneticsoncology.org/Genes/GC_ATXN2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATXN2%5Bgene%5D>
- HGNC Gene Family: Ataxins  
<http://www.genenames.org/cgi-bin/genefamilies/set/411>
- HGNC Gene Family: Trinucleotide repeat containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/775>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=10555](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10555)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6311>
- UniProt  
<http://www.uniprot.org/uniprot/Q99700>

### **Sources for This Summary**

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